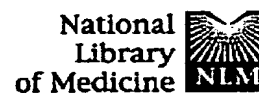


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The human ATP-binding cassette transporter genes: from the bench to the bedside.

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ATP-binding cassette (ABC) transporter genes are ubiquitously present in most organisms from bacteria to man. This gene family is the largest one known as of yet. Still growing, the number of human ABC transporters counts currently 47 members which belong to seven subfamilies. ABC transporters share a similar molecular architecture: (1) Full-structured transporters harbor two symmetric halves each consisting of one nucleotide binding domain (NBD) and one transmembrane domain (TMD). (2) Half-transporters with one NBD and one TMD homo- or heterodimerize to functional transporter complexes. ABC transporters are "traffic ATPases" which hydrolyze ATP and which transport a wide array of molecules or conduct the transport of molecules by stimulating other translocation mechanisms. Many ABC transporters are involved in human inherited or sporadic diseases such as cystic fibrosis, adrenoleukodystrophy, Stargardt's disease, drug-resistant tumors, Dubin-Johnson syndrome, Byler's disease, progressive familial intrahepatic cholestasis, X-linked sideroblastic anemia and ataxia, persistent hyperinsulinemic hypoglycemia of infancy, and others. The present review summarizes the current findings in basic research and the efforts for bridging the gap to clinical applications in therapy and diagnostics.

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